Wisconsin Department of Health Services
Wisconsin Division of Public Health
Newborn Screening Program
Umbrella Committee Meeting
Friday, December 6, 2024
10:00 a.m. - 2:00 p.m.
Zoom: https://dhswi.zoomgov.com/j/1603654162?pwd=VzBzNEZicFZLbm9nN051QWhIcGl5QT09
Meeting ID: 160 365 4162
Or phone: +1 669 254 5252 or +1 646 828 7666 or +1 669 216 1590 or +1 551 285 1373

Minutes

Meeting Members:

X	Dr. Nick Antos		Emily Kittell	X	Leah Ricci
	Jonette Arms	X	Laura Leitch	X	Dr. James Schauer
X	Dr. Mei Baker	X	Sharon Luu	X	Dr. Bob Steiner (Chair)
X	Dr. Donald Basel	X	Dr. Katie Marquart	X	Dr. Julie Thiel
	Anna Benton	X	Dr. Anne Marsh		Angie Thompson
X	Dr. Jeff Britton	X	Dr. Roberto Mendez		Tamara Thompson
	Leah Eckstein	X	Emily Meyer		Paula Tran
X	Dr. Norm Fost	X	Dr. Michelle Miller		Jennifer Ullsvik
X	Dr. John Hokanson		Dr. Pilar Ossorio	X	Isabella Walters
X	Tami Horzewski		Susan Picione	X	Mary Marcus Walters
X	Dr. Julie Kessel	X	Mandy Quainoo		Dr. Jasmine Zapata
X	Alison LaPean-Kirschner				

Meeting Guests:

X	Emilie Blair	X	Zoe Culshaw-Klein		Taylor Duke
X	Sharon Gilbert		Dr. Elizabeth Goetz	X	Dr. Scott-Schwoerer
X	Emma Wagner	X	Kimberly Haugstad	X	Susheela Jayaraman
X	Dean Suhr	X	Wanda Meteer	X	Sara Zoran
X	Bryan Polcyn				

Agenda:

Friday, December 6, 2024 10:00 a.m. – 2:00 p.m.				
Time:	**Topic:**	**Lead:**	**Follow-up Items:**	**Notes:**

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10:00 - 10:10	Welcome Review and Approval of Past Meeting Minutes	Dr. Steiner		Motion to approve May 3, 2024 minutes. 1st motion: Dr. Jeff Britton 2nd motion: Dr. Jamie Schauer Motion approved.
10:10 - 10:20 10:20 – 10:35	Department of Health Services (DHS) Updates WI State Lab of Hygiene (WSLH) Updates	Dr. Thiel/Tami Horzewski Dr. Baker		Dr. Thiel and Tami Horzewski shared the following DHS updates: •Dr. Steiner is back with the NBS Program part-time in the role of DHS NBS Program Medical Director. He works primarily on Tuesdays and Thursdays and every other Friday. • The NBS Program is part of the Family Health Section (FHS). The new FHS Manager, Leah Eckstein, recently started and may join some future meetings. • The Title V, five year needs assessment for setting priorities for maternal and child health work is underway. There were focus groups and a state-wide survey. This information is being analyzed and we are looking at setting

Minutes

				priorities for the next 5 years, 2026-2030. • As mentioned at the previous meeting, DHS will be relocating to a new office/building. The move is now scheduled for next fall, 2025. • Rulemaking for the NBS blood card fee increase and the addition of two conditions to the NBS panel, X-ALD and MPS 1, is moving through the process. The legislative report is in the Governor's Office for review and addressed the comments received during the public hearing/comment period. Rulemaking review will resume when the legislative session begins around January 2025.

				• Infantile Krabbe Disease has been recently added to the Recommended Uniform Screening Panel (RUSP). A small workgroup of metabolic, neurology, and stem cell transplant specialists are currently discussing the work that would need to be done in preparation for possible screening for Infantile Krabbe Disease, including the development of a care infrastructure in state, and the possibility of initial care out of state. There has been discussion of a potential nomination. Dr. Baker provided the following WSLH update: • **X-ALD Demonstration Project Implementation** – Total screened newborns: 59,123

				(9/20/2023 – 9/19/2024) – Reported screen positive: 6 male and 8 female – Confirmed: 5 male (including 1 ZWS) and 5 female – Other outcomes: 2 false positive (1 male and 1 female) - 1 further clinical follow up declined (known carrier mother) - 1 pending (female) - Opt- Out: 20 • **HRSA-23-065: State Newborn Screening Priorities Program (NBS Propel)**

				– **Specific Aim 1:** Expand testing capability to improve laboratory readiness for screening Mucopolysaccharidosis types I and II (MPS I and MPS II), and Guanidinoacetate Methyltransferase (GAMT) deficiency.—The population data evaluation on Guanidinoacetate assay is completed. – **Specific Aim 2:** Improve NBS specimen transit time via increasing transparency and effective communication.—Ongoing – **Specific Aim 3:** Establish a system and a process to monitor

				spinal muscular atrophy screening positive infants and assess treatment efficacy. – It is in progress to establish a REDCap-based 5 year SMA follow-up database and associated dashboard • **The lab received a CF foundation funding of “***Cystic Fibrosis Newborn Screening Sequencing Resource Centers”* . • **CAP Self-Inspection** – The NBS lab underwent a successful Self-CAP inspection on October 30, 2024. No major testing concerns with minor house-keeping issues

				Removing Green Sheet in NBS Cards Working with the Education Subcommittee, a survey was sent out to birth hospitals to determine if the green sheets (inserts providing newborn screening information) should continue to be used. It was determined that the green sheets will no longer be used with the next batch printing in fall of 2025. **2024 APHL NBS Symposium (October 20-24, 2024).** Dr. Baker shared that she provided a presentation on efficient NBS for X-ALD in the section of “Improving Testing Methods and Follow Up in NBS” at the APHL Symposium. Dr. Mendez shared the following information provided at the APHL Symposium: Information on CDC testing programs and standardized lab materials being produced was shared.

				There were discussions on the feasibility of Whole Genome Sequencing (WGS) and NBS – equity for follow up, and storage and protection of data and how AI could be used. There were also discussions on education for families, the importance of inclusion, short term and long term follow up, and information on Lysosomal Storage Diseases, to name a few highlights.
10:35 – 10:45	National Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC)/Recommended Uniform Screening Panel (RUSP) Update	Dr. Steiner		Dr. Steiner shared the following ACHDNC update: Public comment was heard that consisted primarily of advocacy for NBS for Biliary Atresia, Cerebrotendinous Xaanthomatosis (CTX), and Duchenne Muscular Dystrophy (DMD). DMD: nominators have apparently requested a pause in the process, The Metachromatic Leukodystrophy (MLD) nomination was approved for Evidence Review, a brief report was

				given on the progress of the Evidence Review. Infantile Krabbe and Guanidinoacetate Methyltransferase (GAMT) Deficiency are the most recent additions to the Recommended Uniform Screening Panel (RUSP). There was an update on the Laboratory Developed Tests (LDT) —an FDA initiative to regulate them. The LDT issue and presentations on research funding opportunities to document lived experience of patients and Families was shared.
10:45 – 11:00	NBS Research Workgroup Update	Isabella Walters/Dr. Fost		Dr. Fost shared the following Research Workgroup update: The Research Workgroup has been meeting monthly. The group has been discussing the development of policy or guidelines for research using NBS dried blood spots (DBS). The discussion has included the following questions: -who should be involved? -does it need to be an investigator? -does it need to be someone in WI? -what credentials do are needed? -who should be reviewing?

				-should the review go through the committee process and/or a review group at DHS? -how to address issues with consent and opt-in or opt-out mechanisms? -what to do with incidental findings? -how to address issues with IRB review and the steps/process for IRB review? -are there certain qualifications needed for IRB review? The workgroup should be wrapping up and providing their proposed draft guidelines by February. A draft will be sent to the Umbrella Committee and SACNBS for review. It will then be sent to the DHS Secretary for review. Currently there is no research proposal process in place.
11:00 – 11:25	Guanidinoacetate Methyltransferase (GAMT) Deficiency Nomination/Background	Drs. Mendez/Scott-Schwoerer		Dr. Scott-Schwoerer shared background information on GAMT addressing the required review criteria. She provided information on diagnostics, symptoms, natural history, current and recommended treatment, follow-up, management, and outcomes. GAMT is on the

				recommended uniform screening panel (RUSP). Dr. Mendez provided laboratory testing and follow up information. Assay development is in progress.
11:25 – 11:50	GAMT Criteria Review & Voting	Dr. Steiner/Tami Horzewski		The 14 Umbrella Committee voting members reviewed and voted on the nine nomination review criteria. Criteria 1 - Mandated testing should be limited to conditions that cause serious health risks in childhood that are unlikely to be detected and prevented in the absence of newborn screening. Vote: 14 - meets Criteria 2 - For each condition, there should be information about the incidence, morbidity and mortality, and the natural history of the disorder. Vote: 14 - meets Criteria 3 - Conditions identified by newborn screening should be linked with interventions that have been shown in well-designed studies to be

				safe and effective in preventing serious health consequences. Vote: 14 - meets Criteria 4 - The interventions should be reasonably available to affected newborns. Vote: 14 - meets Criteria 5 - Appropriate follow-up should be available for newborns who have a false positive newborn screen. Vote: 11 - meets, 3 more information needed Criteria 6 - The characteristics of mandated tests in the newborn population should be known, including specificity, sensitivity, and predictive value or other convincing. Vote: 14 - meets Criteria 7 – N/A Criteria 8 - Before a test is added to the panel, the details of reporting, follow-up, and management must be

				completely delineated, including development of standard instructions, identification of consultants, and identification of appropriate referral centers throughout the state/region. Vote: 12 – meets, 2 - more info needed Criteria 9 - Recommendations and decisions should include consideration of the costs of the screening test, confirmatory testing, accompanying treatment, counseling, and the consequences of false positives. The mechanism of funding those costs should be identified. Expertise in economic factors should be available to those responsible for recommendations and decisions. Vote: 0 – meets, 1 does not meet, 13- more information needed The Umbrella Committee shared concern regarding criteria #9 and the need for more information to address economic impact. Motion: The Umbrella Committee accepts the metabolic subcommittee

				recommendation to add GAMT to the WI NBS panel **pending additional information regarding economic impact.** - First Motion: Dr. Basel - Second Motion: Dr. Britton Motion approved (10 – yes, 3-abstain, 1-no). There was an additional Motion to table: - First Motion: Dr. Schauer - Second Motion: Dr. Antos Motion **not** approved (3-yes, 10-no, 1-abstain).
11:50 – 12:00	Break			
12:00 – 12:10	Acid Sphingomyelinase Deficiency (ASMD)/Niemann-Pick Disease Nomination/Background	Dr. Steiner		Dr. Steiner provided some background information on ASMD addressing the required review criteria. It was noted as the co-nominator that conflict of interest was disclosed, and the intention was to provide clinical expertise and information for the

				nomination, rather than advocacy for the condition nomination.
12:10 – 12:30	Acid Sphingomyelinase Deficiency (ASMD)/Niemann-Pick Disease Criteria Review & Voting	Dr. Steiner/Tami Horzewski		The 14 voting Umbrella Committee members reviewed the metabolic subcommittee recommendations and nine criteria for the review of the ASMD nomination. Motion: the Umbrella Committee recommendation follows the recommendation of the Metabolic Subcommittee that ASMD should not be added to the WI NBS panel at this time because additional information is needed. • 1st motion: Dr. Britton • 2nd motion: Dr. Hokanson Motion approved. (13-yes, 1-abstain) The Umbrella Committee recommendation will be forwarded on to the Secretary Advisory Committee on Newborn Screening (SACNBS) for the nomination review at the March 14, 2025 meeting.

12:30 – 1:45	Subcommittee Updates Secretary's Advisory Committee on Newborn Screening (SACNBS) Update	All Chairs (CCHD, CF, Education, Endocrine, Hearing, Hemoglobinopathy, Immunodeficiency, and Metabolic) Dr. Fost		**CCHD (Dr. Hokanson)** Dr. Hokanson shared the following update: About 1/1,000 babies fail their screening for various reasons. By updating the blood card more data will be in WE-TRAC so that every baby that needs to be screened is screened. Query has been done looking at just reporting on those babies that need to be screened and not those that already have a known heart condition. During the period of 2014-2022, there were 571, 000 babies in this cohort, 1,000 have a congenital heart diagnosis prenatally and for those diagnosed in a timely fashion before leaving the hospital or 3 days after a homebirth, 42 babies were diagnosed through pulse oximetry. That is approximately 5 babies/year, per the 42 in 9 years. About 102 had late diagnosis with various conditions. The rate of prenatal detection will plateau. The late diagnosis going down, and those identified in a timely fashion postnatally decreased also. Outcomes of babies that failed their echo show 34 had a significant heart disease but didn't classify with the targeted conditions of the screening. A

				small number will have lung disease or various other conditions. There will be a change in screening protocol, updating guidelines from the AAP, changing to only two chances to pass. This may decrease the number of false positives. **CF/Molecular (Dr. Antos)** Dr. Antos shared the following update: CF NBS varies in each state. Wisconsin is one of few states that does next gen sequencing. Information was shared on the new CF Foundation NBS guidelines – looking at an ideal plan. There are seven recommendations. The CF 2023 data summary was shared. There were 15 confirmed CF and CRMS cases (8-CF 7-CRMS) and 6 Spinal Muscular Atrophy (SMA) cases. The CF birth rate is down likely due to prenatal counseling and screening. Information of the testing process and follow up sweat testing was shared.

				There is an increase in listed variants and categorization for different variants. **Education (Alison La-Pean-Kirschner)** Alison La-Pean Kirschner provided the following update: • The Education Subcommittee met most recently on 10/28/24. • Recap of NBS Awareness Month (September 2024) -DHS Facebook posts • Goal of conference attendance and tabling events: -Larger list for next year -this year 4 Kids Health conf -Genetics Exchange -Working toward updates/new stories for the Family Stories page • Subcommittee had worked previously to provide feedback for Family Voices NBS Videos • Educational Materials Work Group (Update) -Current List of Materials -4-pg booklet for Plain Community -Updating 3 screen brochure (blood, CCHD, and Heart)

				-Hearing follow-through postcard -Updating poster and sticker -Working with UW Media solutions for graphics -Updated drafts are expected back 12/18 with an internal working group meeting on 1/2/25 to review. -Redcap survey to elicit feedback when materials are ready for review outside of our subcommittee. -Planning for final versions and printing early in 2025. • Discussed Continued Efforts (and renewed effort moving forward) to engage with NBS education in the prenatal sphere -Specific focus on Community - -Partnerships -Engagement with 2-3 organizations, in-person event opportunities -Possibility of creating a “train the trainer” info package or session for those engaging in community outreach and may have NBS discussion opportunities

				• Vote at Education Subcommittee was unanimous to remove green cards from NBS cards on orders moving forward (based on previous studies of utility, most recent 08/2024) -91 responses from unique birthing units, NICUs or county health departments -59 units reported that they tear out green sheet and provide to families -49 entities do not tear out green sheet, or they do tear it out but store it at the hospital or shred -57 entities (86%) reported that they would NOT be concerned if the green sheet was discontinued -Newborn screening educational materials are currently being updated and should be available by the beginning of 2025. We recommended that organizations replace the green sheet with these educational materials. Concerned respondents were comfortable with this plan.

				-We decided that the parent's copy is not serving the intended purpose and newborn screening education would be more effective through use of the program's one-pagers. **Endocrine (Dr. Marquart)** Dr. Marquart shared the following update: The Endocrine Subcommittee met on November 5. Dr. Mendez shared the 2023 screening summary. There were 63 Congenital Hypothyroidism (CH) cases and 4 Congenital Adrenal Hyperplasia (CAH) cases. The subcommittee is working on adding membership. There has been some turnover with pediatric endocrinologists in the state. The subcommittee is working on adjusting coverage for pediatric endocrinology. There was some discussion on looking at a long term follow up project to see how many patients are still on treatment over time. A population health fellow working with the Milwaukee Health Department attended the meeting and was interested in support services (e.g. coordinating transportation services)

				and education for CH as well as SC patients. **Hearing (Dr. Kessel)** Dr. Kessel Shared the following update: The Hearing Subcommittee discussed screening follow-up and early intervention. information and 2023 quality improvement in hearing screening follow up. 805 babies were rescreened and 549 passed. Goal – to have babies enrolled by six months of age. 51.7% were enrolled and only 33% by six months of age. WI Sound Beginnings (WSB) is working to improve this. 2023 AAP Clinical Report on hearing assessment. Newborn hearing screening is no longer enough. Earlier identification of risk factors for late onset hearing loss was discussed. WSB assessing what is currently happening after newborn screening. AAP guidelines for Congenital Cytomegalovirus (cCMV). Risk factors were discussed for early childhood hearing loss. Treatment recommendations for cCMV in 2024 Red Book.

				DHS has provided grant funding to look at gaps with cCMV and hearing loss. Extended target screening for cCMV. Taskforce and WI stakeholder group - WiSPER: Wisconsin Screening, Prevention and Early Recognition is looking at identifying gaps and tools for screening and education for cCMV. Questions are being discussed on how to track cCMV in Wisconsin. Should this be entered in the WE-TRAC data system? Should this retroactive screening be sent to a testing laboratory? **Hemoglobinopathy (Dr. Marsh)** Dr. Marsh shared the following update: The Hemoglobinopathy Subcommittee met in October. Dr. Baker shared the 2023 screening summary. Hemoglobin trait information was shared. There was a total of 20 newborns identified and confirmed with one of the Hemoglobin (HGB) disorders: 13 sickle cell (SC) disease, 7 HGB SC disease, 1 HGB C-Beta Thalassemia and 1 HGB S-Beta Thalassemia. HBB gene sequencing, assay development, testing, and confirmation were discussed.

				Dr. Singh provided an update on the SC data project work, SC counseling is not a uniform process in Wisconsin. There is a need to look closer at this. There will be more discussion and sharing on this at the next subcommittee meeting. **Immunodeficiency (Dr. Baker)** There is no update as the Immunodeficiency Subcommittee only meets in the spring and the update was provided at the May meeting. **Metabolic (Dr. Basel)** Dr. Basel shared the following update: The Metabolic Subcommittee had a meeting on October 18. Two condition nominations were reviewed and voted on. The Metabolic Subcommittee recommended adding GAMT (currently on the RUSP) to the WI NBS panel. ASMD was also reviewed, and the recommendation was not to add the condition to the panel at this time as more information is needed. Dr. Mendez provided the following 2023 screening summary stating the number of confirmed cases:

				•3 confirmed cases of Galactosemia •6 confirmed cases of late onset Pompe Disease •6 confirmed cases of PKU •8 confirmed Organic Acidurias •2 confirmed Fatty Acid Oxidation Disorders Dr. Baker provided an update on the X-ALD NBS Demonstration project. Six males and eight females were reported as screen positives. A workgroup is looking at Krabbe Disease and how to mobilize rapid transplant that will be required for treatment. The subcommittee will be reviewing MPS 2 at a future meeting.
1:45	Plan Next Meeting/Agenda Items	All		The next meeting will be on Friday, May 2, 2025. • 2024 Screening Summary • Suggested review of false positive data and what is published on adverse effects of false positives in NBS • X-ALD follow up with grant ending • Revisit the role of the Umbrella Committee

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"Parking Lot" Items: